CHARGE Syndrome
For Healthcare Providers

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools (www.gemssforschools.org).

The acronym CHARGE stands for:
C – coloboma
H – heart abnormalities
A – atresia choanae
R - retarded growth and development
G – genital abnormalities
E – ear anomalies

Physical characteristics and/or symptoms

Note: not all people with CHARGE syndrome will have all of these features.

Common Characteristics

- Ocular abnormalities (80-90%)
  - Unilateral or bilateral coloboma of the iris, retina-choroid, optic nerve
  - Microphthalmos
- Choanal atresia or stenosis (50-60%)
  - Unilateral or bilateral
  - May be obscured with cleft lip/palate
- Cranial nerve dysfunction
  - Decreased or absent sense of smell
  - Unilateral or bilateral facial palsy (40%)
  - Hearing loss
  - Swallowing problems (70%-90%)
- Ear anomalies (80-100%)
  - Outer ear anomalies
    - Often a typical "CHARGE" ear that is short and wide with small lobes
    - Inner ear folds are prominent and the outer folds appear “snipped off” (for an illustration, go to this link: http://www.ncbi.nlm.nih.gov/books/NBK1117/figure/charge.F1/?report=objectonly
  - Middle ear anomalies (common)
    - Bones of the middle ear are malformed

        - Causes conductive hearing loss
        - Prevalence of severe to profound hearing loss is 50%
    - Inner ear anomalies
        - Malformed
- Cryptorchidism in males and hypogonadotropic hypogonadism in both males and females
- Developmental delay
    - Marked delays in motor development
    - Speech delays often related to hearing loss of clefting
    - Eventual cognitive abilities often normal
    - Can have behavioral problems that include ADHD, self-injurious behaviors or obsessive-compulsive symptoms
- Cardiovascular malformations (75%-85%)
- Growth deficiency (70%-80%)
- Orofacial clefts (15%-20%)
- Tracheoesophageal fistula (15%-20%)
- Characteristic facial features (in addition to the outer ear anomalies)
    - Square face
    - Broad prominent forehead
    - Arched eyebrows
    - Large eyes
    - Occasional droopy lids
    - Prominent nasal bridge and thick nostrils
    - Prominent nasal columella
    - Flat midface
    - Small mouth
    - Small chin
    - Facial asymmetry with or without facial palsy

Recommended Routine Surveillance

- Assessment of feeding/ swallowing difficulties
- Cardiac evaluation
- Coanal atresia
- Ophthalmologic and auditory evaluation
- Surveillance for scoliosis
- Routine Dental care

Emergency Protocols

There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation.

- Emergencies should be handled as with any child.

- If seizures are present, the following seizure action plan may be useful: https://www.aap.org/en-us/Documents/Seizure_Action_Plan_for%20School.pdf

Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Cardiologist
  - Surveillance for cardiac defects
- Dentist:
  - Microdontia
  - Enamel hypoplasia
  - Malocclusion
- Developmental evaluation:
  - Speech therapy
  - Physical therapy
  - Occupational therapy
  - Deaf/blind specialist
- ENT
  - Choanal atresia
  - Hearing loss
  - Ear anomalies
- Endocrinologist
  - Growth deficiencies
  - Hypogonadotropic hypogonadism
- Gastroenterologist
  - Significant feeding problems may occur
  - Gastrointestinal reflux
  - Swallowing difficulties Orthopedist:
  - Scoliosis
  - Limb/bone abnormalities
- Geneticist / Genetic Counselor
  - Diagnosis
  - Coordination of care
  - Genetic risk for family
  - Clinical trials
- Nephrologist
  - Renal anomalies
- Neurologist
  - Brain abnormalities
- Ophthalmologist
  - Coloboma

    - Microphthalmos
    - Cataracts (adults)
- Orthopedist
    - Scoliosis
    - Limb/bone abnormalities
- Pulmonologist
    - Breathing difficulties
- Urologist
    - Genital abnormalities

Sample Forms

- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

My patient_____ has been diagnosed with CHARGE syndrome. CHARGE Is a mnemonic for coloboma, heart defects, choanal atresia, retarded growth and developmental, genital abnormalities, and ear anomalies. Medical complications with CHARGE syndrome include management of feeding difficulties, heart defects, choanal atresia, hearing loss, visual defects, scoliosis, gastrointestinal reflux, and constipation. Because of these, _____ needs the following accommodations.

Seven Important Aspects of School Life

"CHARGE Syndrome at a Glance" will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions

Resources

Gene Reviews
http://www.ncbi.nlm.nih.gov/books/NBK1117/

Genetic Home Reference
https://ghr.nlm.nih.gov/condition/charge-syndrome